

Clinical utility of genomics in perinatal autopsy

Background

Melbourne Genomics' 16 Clinical Flagships have been at the forefront of determining when genomic testing makes a demonstrable difference to the safety and quality of patient care.

In Victoria, a state-wide service investigates the deaths of babies who die during pregnancy or early infancy (the Victorian Perinatal Autopsy Service, VPAS).

Parents' distress at the loss of a baby is often exacerbated by not knowing the exact cause, as well as uncertainty about the chance of the same condition occurring in future pregnancies.

Project description

The objective: to determine if genomic sequencing can be incorporated into post-mortem examination by skilled pathologists, and whether this process results in more parents getting an answer about the cause of their baby's condition.

The project also investigated whether being able to offer sophisticated genomic testing during pregnancy could have been helpful in finding the cause of an infant's condition before birth.

The Perinatal Autopsy Flagship was led by Dr George McGillivray and Dr Jackie Collett (both from The Royal Children's Hospital). Flagship clinician Dr Fiona Chan provided key coordination, with at least 24 medical professionals were directly involved.

Activities, outcomes and lessons learnt

Findings from this project will be made available following publication of results.

Clinical Flagship team

Name	Organisation	Role
George McGillivray	RCH/MCRI	Clinical geneticist
Jacqueline Collett	RCH	Anatomical pathologist
Fiona Chan	RCH	Anatomical pathologist
Alison Yeung	Monash Health/RCH/MCRI	Clinical geneticist
Anand Vasudevan	UoM/RWH	Clinical geneticist
Anna Jarmolowicz	MCRI/VCGS	Genetic counsellor
Beena Kumar	Monash Health	Anatomical pathologist
Elske Pasma	UoM/RWH	Maternal foetal medicine obstetrician
Gemma Brett	MCRI/VCGS	Genetic counsellor
Giulia Valente	Austin Health	Genetic counsellor
Heather Chaliner	Austin Health	Genetic counsellor
Ian Simpson	Monash Health	Anatomical pathologist
Jan Pyman	RCH/RWH	Anatomical pathologist
Kerryn Ireland-Jenkin	Austin Health	Pathologist
Mark Teoh	Monash Health	Foetal diagnostic ultrasound specialist and obstetrician
Matthew Hunter	Monash Health	Clinical geneticist
Natasha Brown	MCRI/VCGS	Clinical geneticist
Sam Ayres	MCRI/VCGS	Genetic counsellor
Sue Walker	UoM	Maternal foetal medicine obstetrician
Susan White	RCH/MCRI	Clinical geneticist
Trishe Leong	Austin Health	Anatomical pathologist
Yael Praver	Monash Health	Genetic counsellor
Yuen Chan	Monash Health	Pathologist
Zornitza Stark	MCRI/VCGS	Clinical geneticist